KCNE1 gene

potassium voltage-gated channel subfamily E regulatory subunit 1

Normal Function

The *KCNE1* gene provides instructions for making a protein that regulates the activity of potassium channels. These channels, which transport positively charged potassium atoms (ions) into and out of cells, play a key role in a cell's ability to generate and transmit electrical signals.

The specific function of a potassium channel depends on its protein components and its location in the body. The KCNE1 protein regulates a channel made up of proteins produced by the *KCNQ1* gene. Four alpha subunits, each made from the *KCNQ1* gene, form the structure of each channel. One beta subunit, produced from the *KCNE1* gene, binds to the channel and regulates its activity.

These channels are active in the inner ear and in heart (cardiac) muscle, where they transport potassium ions out of cells. In the inner ear, the channels play a role in maintaining the proper ion balance needed for normal hearing. In cardiac muscle, the channels are involved in recharging the muscle after each contraction to maintain a regular heartbeat. The KCNE1 protein is also produced in the kidneys, testes, and uterus, where it probably regulates the activity of other channels.

Health Conditions Related to Genetic Changes

Jervell and Lange-Nielsen syndrome

Several mutations in the *KCNE1* gene have been identified in people with Jervell and Lange-Nielsen syndrome. Affected people typically have mutations in both copies of the *KCNE1* gene in each cell. These mutations change a single protein building block (amino acid) in the KCNE1 protein, which disrupts the protein's normal structure. An altered KCNE1 protein cannot regulate the flow of potassium ions through channels in the inner ear and cardiac muscle. This loss of channel function leads to the profound hearing loss and abnormal heart rhythm (arrhythmia) characteristic of Jervell and Lange-Nielsen syndrome.

Romano-Ward syndrome

More than 30 *KCNE1* mutations are known to cause Romano-Ward syndrome. Usually, people with this condition have a mutation in only one copy of the *KCNE1* gene in each cell. These mutations change a single amino acid in the KCNE1 protein, which alters the protein's ability to regulate potassium channels in cardiac muscle

cells. A disruption in the flow of potassium ions out of these cells causes the irregular heartbeat found in Romano-Ward syndrome.

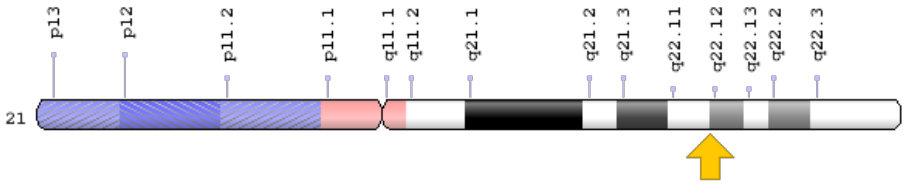
other disorders

Certain drugs, including medications used to treat arrhythmias, infections, seizures, and psychotic disorders, can lead to an abnormal heart rhythm in some people. This drug-induced heart condition, which is known as acquired long QT syndrome, increases the risk of cardiac arrest and sudden death. A small percentage of cases of acquired long QT syndrome occur in people who have an underlying mutation in the *KCNE1* gene.

Chromosomal Location

Cytogenetic Location: 21q22.12, which is the long (q) arm of chromosome 21 at position 22.12

Molecular Location: base pairs 34,446,688 to 34,512,275 on chromosome 21 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- Delayed rectifier potassium channel subunit IsK
- IKs producing slow voltage-gated potassium channel beta subunit Mink
- ISK
- JLNS2
- KCNE1_HUMAN
- LQT5
- Minimal potassium channel
- minK
- potassium channel, voltage gated subfamily E regulatory beta subunit 1
- potassium voltage-gated channel, Isk-related family, member 1

Additional Information & Resources

GeneReviews

- Jervell and Lange-Nielsen Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1405
- Long QT Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1129

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28KCNE1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- POTASSIUM CHANNEL, VOLTAGE-GATED, ISK-RELATED SUBFAMILY, MEMBER 1
  http://omim.org/entry/176261

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_KCNE1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=KCNE1%5Bgene%5D
- HGNC Gene Family: Potassium voltage-gated channel regulatory subunits
  http://www.genenames.org/cgi-bin/genefamilies/set/858
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=6240
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/3753
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